### **PCT**

## RLD INTELLECTUAL PROPERTY ORGANIZATION International Bureau



# INTERNATIONAL APPLICATION PUBLISHED UNDER THE PATENT COOPERATION TREATY (PCT)

(51) International Patent Classification 6:
C12Q 1/68

A1

(11) International Publication Number: WO 96/02671

(43) International Publication Date: 1 February 1996 (01.02.96)

(21) International Application Number:

PCT/SE95/00804

(22) International Filing/Date:

9403953-4

29 June 1995 (29.06.95)

(30) Priority Data: 9402487-4 \

15 July 1994 (15.07.94) SE 16 November 1994 (16.11.94) SE

(71) Applicant (for all designated States except US): PHARMACIA BIOTECH AB [SE/SE]; S-751 82 Uppsala (SE).

(72) Inventors; and

- (75) Inventors/Applicants (for US only): BYWATER, Margaret [GB/SE]; Stigbergsvägen 11, S-752 42 Uppsala (SE). LINDSTRÖM, Per [SE/SE]; Svankärrsvägen 26A, S-756 53 Uppsala (SE). INGANÄS, Mats [SE/SE]; Lapplandsresan 14, S-757 55 Uppsala (SE).
- (74) Agents: EBBINGHAUS, Marie-Louise et al.; Pharmacia AB, Patent Dept., S-751 82 Uppsala (SE).

(81) Designated States: JP, US, European patent (AT, BE, CH, DE, DK, ES, FR, GB, GR, IE, IT, LU, MC, NL, PT, SE).

### Published

With international search report.

Before the expiration of the time limit for amending the claims and to be republished in the event of the receipt of amendments.

(54) Title: SEQUENCE-BASED MUTATION ANALYSIS OF NEOPLASTIC TISSUE FOR DIAGNOSIS OR PROGNOSIS OF THE NEOPLASIA

#### (57) Abstract

A method for sequence-based diagnosis of a human neoplastic tissue, blood or other body fluid sample, comprises analysing from genomic DNA or cDNA derived from said neoplasia the DNA sequence of a gene encoding a cancer-related protein for the presence of mutations therein, determining from the presence, nature and location of any such mutation or mutations the influence thereof on the biological function of the corresponding protein and thereby on the properties of the neoplasia, and on the basis thereof prognosticating the development of the neoplasia.